

What is claimed is:

1. An isolated or recombinant nucleic acid associated with the presence of multiple endocrine neoplasia type 1, wherein said nucleic acid encodes a protein defined as follows:
- (i) having a calculated molecular weight of about 67.5 kDa; and
 - (ii) (a) specifically binding to an antibody raised against an protein with a sequence as set forth in SEQ ID NO:2; or
 - (b) having at least 60% amino acid sequence identity to a protein with a sequence as set forth in SEQ ID NO:2.
2. The isolated or recombinant nucleic acid of claim 1, which further comprises non-coding sequence.
3. The isolated or recombinant nucleic acid of claim 2, wherein the non-coding sequence comprises introns.
4. The isolated or recombinant nucleic acid of claim 3, wherein the sequence is SEQ ID NO:3.
5. The isolated or recombinant nucleic acid of claim 1, wherein the nucleic acid sequence encodes a protein having at least 80% amino acid sequence identity to a protein with a sequence as set forth in SEQ ID NO:2.
6. The isolated or recombinant nucleic acid of claim 5, wherein the nucleic acid sequence encodes a menin protein with a sequence as set forth in SEQ ID NO:2.
7. The isolated or recombinant nucleic acid of claim 1, wherein the sequence is SEQ ID NO:1.
8. The isolated or recombinant nucleic acid of claim 1, wherein the nucleic acid sequence specifically hybridizes to SEQ ID NO:1 under stringent conditions.

9. An isolated or recombinant protein defined as follows:
- (i) having a calculated molecular weight of about 67.5 kDa; and
 - (ii) (a) specifically binding to an antibody raised against an protein with a sequence as set forth in SEQ ID NO:2; or
 - (b) having at least 60% amino acid sequence identity to a protein with a sequence as set forth in SEQ ID NO:2.
10. The isolated or recombinant protein of claim 9, wherein the protein has at least 80% amino acid sequence identity to a protein with a sequence as set forth in SEQ ID NO:2.
11. The isolated or recombinant protein of claim 10, wherein the protein has sequence as set forth in SEQ ID NO:2.
12. An antibody, specifically immunoreactive under immunologically reactive conditions, to a protein comprising SEQ ID NO:2.
13. An antibody, specifically immunoreactive under immunologically reactive conditions, to a protein, wherein the protein is encoded by a nucleic acid,
- wherein the nucleic acid encodes a protein defined as having a calculated molecular weight of about 67.5 kDa; and (a) specifically binding to an antibody raised against an protein with a sequence as set forth in SEQ ID NO:2; or (b) having at least 60% amino acid sequence identity to a protein with a sequence as set forth in SEQ ID NO:2.
14. A method for detecting the presence of menin in a human cell or tissue, said method comprising:
- (i) isolating a biological sample from a human being tested for menin;
 - (ii) contacting the biological sample with a menin specific reagent; and,
 - (iii) detecting the level of menin specific reagent that selectively associates with the sample.

15. The method of claim 14, wherein the menin specific reagent is selected from the group comprising: menin specific antibodies, *MEN1* amplification primers and nucleic acid probes which selectively bind to *MEN1*.

16. The method of claim 14, wherein the contacting step uses a menin specific antibody.

17. The method of claim 14, wherein the human from which the sample is isolated is suspected of being at risk from multiple endocrine neoplasia type 1.

18. The method of claim 14, wherein the contacting step uses a *MEN1* specific PCR primer pair that amplifies a region of the *MEN1* gene in which a mutation has been associated with multiple endocrine neoplasia type 1.

19. A method for detecting in a test sample the presence or absence of a mutation in a nucleotide sequence essentially encoding human menin or the presence or absence of *MEN1* allele comprising;

a) contacting said test sample suspected of containing a gene missing a *MEN1* allele or encoding a mutant form of the human menin with a first oligonucleotide having a sequence competent to discriminate between the wild type gene and the missing allele or mutant form; and,

b) detecting the formation of a duplex between the gene and the first oligonucleotide sequence.

20. A method of claim 19, wherein the first oligonucleotide is unable to bind to the wild-type *MEN1* gene under hybridization conditions in which the first nucleotide binds to the mutant sequence of *MEN1*.

21. A method of claim 19, wherein the contacting step further comprises amplifying a portion of the human *MEN1* gene and where the first nucleic acid is a polymerase chain reaction amplification primer which binds to an intron of *MEN1*.

22. A method of claim 19, wherein the contacting step further comprises amplifying a portion of *MEN1* and where the first nucleic acid is a polymerase chain reaction amplification primer which discriminates between wild-type and mutant forms of *MEN1* using allelic specific polymerase chain reaction.

23. A method of claim 19, wherein the first nucleic acid binds to either exons or introns of the genomic DNA encoding the human menin gene.

24. A kit for detecting in a test sample the presence or absence of a mutation in a nucleotide sequence corresponding to the wild type allele encoding menin comprising;

a) a container holding a first oligonucleotide sequence whereby said first nucleotide sequence is capable of discriminating between the wild type gene and the mutant form; and

b) a container holding a reagent for detecting the formation of a duplex between the gene and the first nucleotide sequence.

25. A kit for detecting in a test sample the presence or absence of a mutation in a nucleotide sequence essentially encoding menin comprising;

a) a container holding a first nucleotide sequence whereby said first nucleotide sequence is capable of discriminating between the wild type gene and the mutant form; and

b) a container holding a reagent for detecting the formation of a duplex between the gene and the first nucleotide sequence.

26. The kit of claim 25, further comprising amplification primer pairs specifically binding to a human genomic DNA sequence containing *MEN1*.

27. A kit for detecting the presence or absence of menin in persons at risk for multiple endocrine neoplasia type 1 comprising a first container containing an antibody specific for menin and a second container containing an antigen that specifically binds to the menin specific antibody in the first container.

28. The kit of claim 25, wherein the antibody can detect the presence of a wild-type menin protein.

29. The kit of claim 25, wherein the antibody can detect the presence of a mutated menin protein.

Sub D9 30. A transfected cell comprising a heterologous nucleic acid encoding a menin protein or subsequence thereof.

31. A transfected cell into which an exogenous nucleic acid sequence has been introduced, the exogenous nucleic acid specifically hybridizing under stringent conditions to a nucleic acid with:

a nucleic acid sequence as set forth in SEQ ID NO:1 or SEQ ID NO:3; or,
a nucleic acid encoding a protein defined as having a calculated molecular weight of about 67.5 kDa; and (a) specifically binding to an antibody raised against an protein with a sequence as set forth in SEQ ID NO:2; or (b) having at least 60% amino acid sequence identity to a protein with a sequence as set forth in SEQ ID NO:2; and,
the cell expresses the exogenous nucleic acid as a menin protein.

a 32. The transfected cell of claim 30 or 31, wherein the heterologous or exogenous nucleic acid comprises a nucleic acid as set forth in SEQ ID NO:1 or SEQ ID NO:3.

a 33. The transfected cell of claim 30 or 31, wherein the cell is a human cell.

34. An organism into which an exogenous nucleic acid sequence has been introduced, the exogenous nucleic acid specifically hybridizing under stringent conditions to a nucleic acid with:

a sequence as set forth in SEQ ID NO:1; or,

a nucleic acid encoding a protein defined as having a calculated molecular weight of about 67.5 kDa; and (a) specifically binding to an antibody raised against an protein with a sequence as set forth in SEQ ID NO:2; or (b) having at least 60% amino acid sequence identity to a protein with a sequence as set forth in SEQ ID NO:2; and, the organism expresses the exogenous nucleic acid as a menin protein.

35. The organism of claim 34, wherein the exogenous nucleic acid comprises the nucleic acid as set forth in SEQ ID NO:1 or SEQ ID NO:3.

36. An expression cassette comprising a nucleic acid encoding a menin polypeptide, wherein the nucleic acid is operably linked to a promoter and comprises a nucleic acid encoding a menin polypeptide.

37. The expression cassette of claim 36, further comprising an expression vector.